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Newborn screening for congenital hypothyroidism and phenylketonuria—Beyond cost savings

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To the Editor:

Although we agree with Appelberg et al that newborn screening (NBS) for phenylketonuria (PKU) and congenital hypothyroidism (CH) is of great value,¹ we disagree that such results in net cost-savings. Their contention relies on assumptions derived from cost-benefit studies published 4 decades ago.^{2,3} In 2005, a commentary in this journal challenged those arguments.⁴ We later reviewed the frequencies of intellectual disability among individuals with late-treated PKU and CH^{5,6}; the latter review is the second reference cited in new CH clinical guidance.⁷ Our subsequent reviews of economic evaluations of NBS for PKU and CH pointed out that unrealistic assumptions about late-treated CH and PKU, like those made by Appelberg et al, result in overestimates of economic benefits.^{8–10}

The authors state that their estimates of the probabilities of intellectual disability with and without screening for CH and PKU “used published literature.” Of the five cited references, one refers to unpublished clinical data, one is a book chapter,¹¹ and three are economic assessments that cited case series from the 1960s.^{2,3,12} Appelberg et al elsewhere cited our two economic critiques,^{9,10} acknowledging, “The outcome of no screening may be too pessimistic, not reflecting today’s healthcare system, leading to an overestimation of the benefits of screening, making it difficult to determine results to be cost-saving (47, 48).” In their defense, they assert “the lack of data regarding the no screening alternative, due to the fact that the screening program has existed for so long that no untreated affected individuals exist in Sweden.” We disagree; our articles cited published data on outcomes for Swedish children with CH born before the newborn screening.¹³

Optimizing child development is sufficient to justify extending CH screening and treatment to the 70% of the world’s children who lack access to NBS.¹⁴ There is no need to overestimate cost savings from NBS for CH.

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